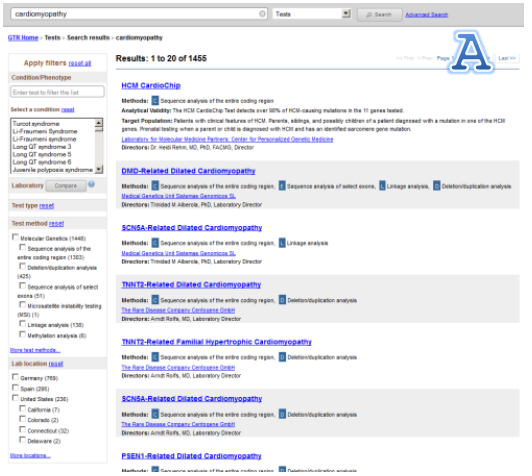
Introducing

GENETIC TESTING REGISTRY

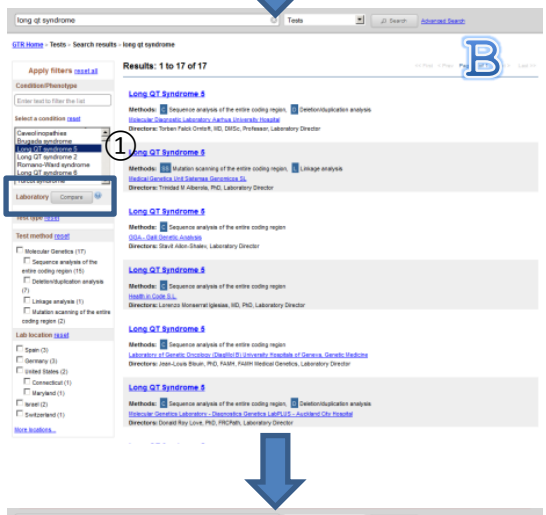
http://www.ncbi.nlm.nih.gov/gtr/

What is the GTR?

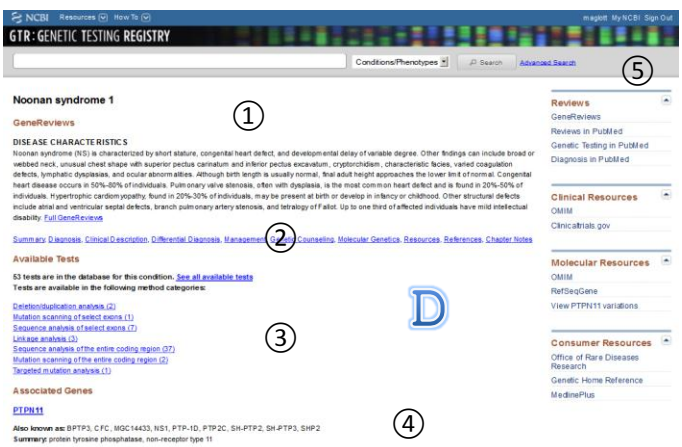
The NIH Genetic Testing Registry (GTR) is a centralized repository of genetic test information voluntarily supplied by test providers. The scope of the Registry includes the purpose, methodology, validity, and evidence of utility of each test, as well as laboratory contacts and credentials. The web site links to context-specific information about conditions, genes, test standards, practice guidelines, and consumer support sites.



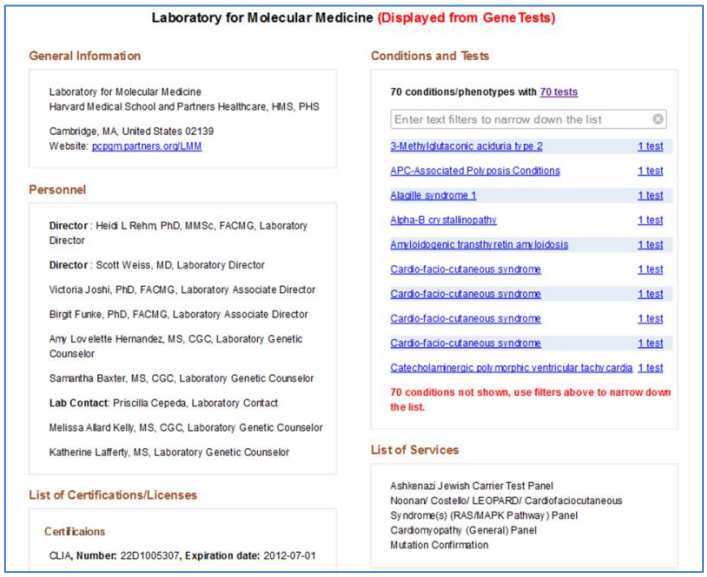
The GTR is a test registry, not a laboratory directory. The GTR enables centralized access to detailed information about genetic tests by collecting sufficient information to document the basis and intended purpose(s) of the test, exactly what is measured and how that measurement is done, and the evidence that supports the test's analytical validity, clinical validity, and clinical utility. The web interface allows users to retrieve that information from multiple avenues, including by name of the test, by name of the provider, by category of the test, and by purpose (*e.g.,* name of condition or gene). If a user does a non-specific search (**A**) for tests (note the menu selection in the query bar present on all pages), and many results are found, the filters in the left column can be used to make selections and quickly focus on the subset of the results of interest. For example, the 1455 results for a query of cardiomyopathy in **(A)** would quickly be reduced to 17 if Long QT Syndrome 5 were checked in the Apply filters section (**B.1**). When one disease is selected, then the Laboratory Compare button (boxed in **B**) can be used to generate a grid (**C**) summarizing the categories of available tests for that condition from each test provider. Each provider-specific section can be expanded by the control at the right to display more specifics about each category of test (**C.1**).



The web site provides a portal for medical geneticists.
The GTR web site provides multiple views of medical genetic information: Test details, laboratory summaries, condition-specific reports, and gene-specific reports. This approach makes it easy to display key information, and quickly navigate to the specifics of choice. For example, in **(D)**, which is a page specific to a condition, you see the disease characteristics portion of a *GeneReviews* summary (**D.1**), with links to the complete *GeneReviews* and each section (**D.2**); a summary of the tests available (**D.3**), with links to more details about methodologies; a list of associated genes (**D.4**); and context-specific links to review articles, practice guidelines, and resources for clinical practice, review of molecular biological details, and consumer resources (**D.5**).



Your laboratory in the GTR

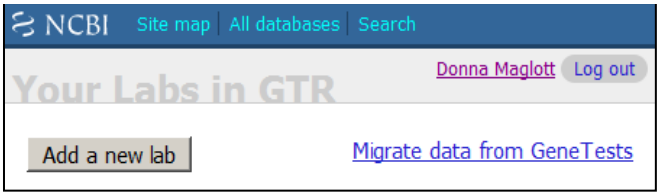


Laboratory-specific pages in the GTR display the tests and services offered, as well as how to contact the laboratory. This information is provided by the laboratory.

Laboratories choose which staff members they want to show publically in the GTR.

Users can bookmark their favorite labs for easy access to the lab's test menu and contact information.

Users can browse the conditions/phenotypes and the associated tests offered by a single laboratory, and view the additional services a laboratory performs.



Submit test information to the GTR

- Transfer data from GeneTests in one simple click!
- Add, edit and delete your tests online.
- Use GTR's specialized web interface to facilitate submission of supplemental data.
- Take advantage of the bulk upload feature via spreadsheet or XML.

The GTR utilizes the latest software and internet technologies for searching, registration and submission, and provides a range of tools to simplify and speed the process of registering tests. The data entry system has been designed to minimize burden to registrants, with extensive use of menus, "type ahead" functionality, and text fields for those components where submitters might want to cut-and-paste information from their websites and other sources. Where possible, fields are automatically populated for the submitter; for instance, once a submitter fills out the condition for which a test is used, several related fields (*e.g.*, disease identifiers, synonyms, acronyms and disease types) are automatically populated for review. A test copy function is available to expedite entry of information for different tests for which many fields are the same*, e.g.*, if the new test differs in only a few fields such as test name and order code. Submitters can then change any field from the copied test. The GTR also provides a mechanism for bulk submission of data, which will significantly reduce the burden for laboratories that want to provide information on multiple genetic tests or multiplex panel tests.

What information is required?

The GTR is intended to provide information about the test provider as well as the availability, accuracy, validity and usefulness of each test. The complete list of optional (85) and minimal (31) fields being requested is posted on the GTR web site at http://www.ncbi.nlm.nih.gov/projects/gtr/docs/GTR_Proposed_Field_Definitions_distribute.pdf. About half of the minimal fields describe the laboratory (*e.g.,* name, contacts), and are automatically completed after the first entry into the GTR. If a test is migrated from GeneTests, approximately 26 fields are transferred and automatically populated in GTR, leaving only 5 fields to add. For tests being submitted *de novo*, minimal fields are clearly marked for easy entry.

Contact us:

Stay informed of the latest GTR news! Email us at gtr@ncbi.nlm.nih.gov and ask to be subscribed to the GTR listserv. GTR is projected to launch in early 2012. Bookmark NOW! http://ncbi.nlm.nih.gov/gtr